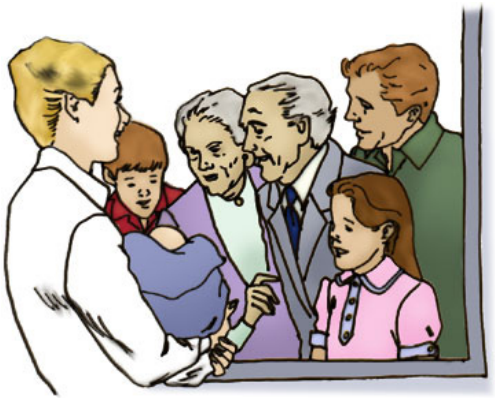
X-Plain™
Newborn Screening Tests
Reference Summary

Introduction

Newborn Screening is a simple blood test used to identify many life-threatening diseases before symptoms begin. Even though each disease by itself is rare, all together these diseases affect 1 in 1500 babies. If untreated they can lead to growth problems, mental retardation or even death.



Newborn screening usually consists of a simple blood test that involves pricking the baby's heel to get a few drops of blood. Multiple tests are run on the blood and if those results are not normal, the lab notifies the baby's doctor. Once diagnosed, treatment can save the baby from death or lifelong health problems.

This health education summary explains newborn screening. It discusses its reasons, the procedure and interpreting results.

Why Screen

Newborn Screening is important because a baby with one of these illnesses may seem healthy but by the time symptoms appear, permanent damage may have occurred. Damage can lead to mental retardation or, in many cases, death.

With the advances in medical technology, it is now possible to find extremely small amounts of body chemicals in the blood. This shows whether a newborn baby is likely to have one of these diseases. Additional laboratory testing, such as genetic testing, can confirm if the newborn has some of these diseases.

Some of these newborn diseases are very dangerous. One example is a rare disease called MCAD. Twenty five percent of children with MCAD die with their first illness. The treatment for MCAD can be as simple as avoiding fasting and giving the child frequent meals.

Phenylketonuria, or PKU, was the first disease to be screened for in newborns. People with PKU do not have the enzyme that processes phenylalanine (Phe). Because of this, phenylalanine builds up in the blood and brain, which leads to mental retardation. Treatment for PKU simply involves a special diet with very small amounts of Phe. This allows the child to grow normally without mental or physical retardation.

Screening newborns for such dangerous and rare diseases gives them the best chance for a healthy life. Even though all states require a newborn screening, each state screens for different disorders. A newborn screening can test for more than 50 disorders, but most states screen for less than 8. It is a parent's responsibility to find out how many disorders their state screens for.

Procedure

The newborn screening test is very simple. The test is performed by pricking your baby's heel and putting a few drops of blood on a special filter paper. The paper is allowed to dry and then sent to a laboratory, where several different tests will be performed.

It is better if the sample is taken when a baby is between 24 hours and 7 days old. This is because some diseases, such as PKU, do not always show up if the blood sample is taken too soon after birth. If the mother and newborn are discharged before the baby is 24 hours old, the test should be done before they leave and repeated 1 to 2 weeks later.

Results

Some labs send the results to the doctor and others send them to the hospital where the baby was born. Ask your doctor how you will get the results and when you should expect them.

If a test result comes back "normal," it means there is no indication that the baby has one of the rare diseases. If a test result comes back "abnormal," it either means the baby may have one of the rare diseases tested for or that the test result is a "false positive." Either way, more testing is needed.

If further tests confirm that your child has a disorder, your child's doctor may refer you to a specialist for treatment. If you have other children who were not screened for the disorder, you may want to have them screened. They may be at risk for the disorder and need treatment also.

Risks

There are no complications involved with pricking a baby's heel for a blood sample.

A test that shows an abnormality is called a "positive" test. If a screening test comes back positive but further tests are negative, it is called a "false positive." The only risk involved with a false positive test is the anxiety it might cause parents while waiting for final test results.

State Requirements

Most states, the District of Columbia, Puerto Rico, and the U.S. Virgin Islands have mandatory newborn screening programs. However, these programs differ. Some states only screen for 4 disorders while others screen for more than 30 disorders. In Wyoming and Maryland, the screening is not mandatory.

A lot of states offer expanded screening with tandem mass spectrometry[1] on every baby. Some hospitals offer screening beyond what state law requires. If your hospital does not offer expanded screening and your doctors think it would be worthwhile, you may want to find a laboratory that provides supplemental testing.

Additional Tests

Additional screening can be for

- genetic disorders such as muscular dystrophy
- infections such as HIV and
- different kinds of cancer such as neuroblastoma

These are disorders that are not usually tested for in required state screening.

The decision whether or not to get additional screening can be a difficult one. You should discuss this with your doctor. If your baby has a close relative who has an inherited disorder or who has died from one, you should consider additional screening.

If you have health insurance, keep in mind that it may not pay for additional screening. You may have to pay it out of your own pocket. This should NOT change your decision to get additional screening. If you will have trouble paying for additional screening,

make sure to let your doctor know. She or he will refer you to a social service specialist who can probably help you find financial help.

Screened Disorders

There are several types of diseases that can be found through newborn screening, such as metabolic disorders, hormonal disorders, blood disorders and hearing disorders. Most of the required newborn screening tests are for metabolic disorders.

Metabolic disorders have to do with metabolism, which is the way the body uses nutrients to keep tissues healthy and produce energy. Hormonal disorders have to do with hormones, which are chemicals in the blood that regulate important bodily functions.

Screening newborn hearing is required in many states. It is possible to screen a baby's hearing before he or she leaves the hospital. If your baby's hearing is not checked at birth, make sure he or she gets tested before turning 3 months old. Diagnosing hearing problems early makes a big difference, since early treatment improves the long term outcome. Unrecognized hearing loss can cause learning disabilities.

The following are examples of metabolic disorders.

- PKU[2]
- Galactosemia[3]
- Biotinidase deficiency[4]
- MSUD[5]
- Homocystinuria[6]

The following are examples of hormonal disorders. Click on a term to learn more about it.

- Congenital hypothyroidism[7]
- Congenital adrenal hyperplasia[8]
- Sickle cell anemia[9] is an example of a blood disorder.

Conclusion

Newborn screening is a blood test done on newborn babies to find out if they have certain harmful or fatal diseases that are not obvious at birth but can be treated. Such diseases are usually rare. However, they hinder an infant's normal development in a variety of ways, physically and mentally.

Newborn screening usually consists of a simple blood test that involves pricking the baby's heel to get few drops of blood. The lab results tell doctors and parents whether newborns have certain conditions that could eventually cause problems. Once diagnosed, treatment can save the baby from lifelong health and developmental problems or even death.

[1] A new technology called tandem mass spectrometry or MS/MS screens for more than 20 disorders with a single test. Because of this technology, more states are beginning to offer expanded newborn screening tests.

[2] Babies affected with PKU cannot process an amino acid, which results in mental retardation. This condition is treated with a special diet.

[3] Babies that have galactosemia lack the enzyme that converts galactose into glucose. Galactosemia can lead to liver failure, cataracts, severe mental retardation and even death. Following a special diet treats the condition.

[4] Babies who have biotinidase deficiency don't have enough of the enzyme biotinidase. This condition causes seizures, poor muscle control, immune system impairment, hearing loss, mental retardation, coma, and even death. Giving the baby biotin treats this condition.

[5] Babies affected with MSUD cannot process multiple amino acids. If not detected and treated early, MSUD can cause mental retardation, physical disability and even death. The condition can be controlled with a special diet.

[6] Babies with homocystinuria cannot break down homocystine. This condition can lead to dislocated lenses of the eyes, mental retardation, skeletal abnormalities and abnormal blood clotting. The disorder can be controlled through a special diet and medications.

[7] Babies that have congenital hypothyroidism don't have enough thyroid hormone, which causes physical and mental retardation. Giving the baby thyroid hormone treats this condition.

[8] Babies that have congenital adrenal hyperplasia lack certain hormones produced by the adrenal gland. This condition can affect the development of the genitals and may cause death due to loss of salt from the kidneys. It can be managed by giving the baby needed hormones.

[9] Babies with sickle cell anemia have red blood cells that form abnormal "sickle" shapes, which can cause episodes of pain, damage to vital organs, strokes, infections and even death. Complications of this condition can be managed with preventative antibiotics and close monitoring of blood counts.